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ACMG STATEMENT

Stewardship of patient genomic data: A policy statement of the American College of Medical Genetics and Genomics (ACMG)

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Disclaimer: This statement is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this statement is completely voluntary and does not necessarily assure a successful medical outcome. This statement should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.
Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this statement. Clinicians also are advised to take notice of the date this statement was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

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Human genomic data linked to health records have become valuable in the quest to establish correlations between disease and genetic information. As a result, it has become increasingly common for patient genetic information obtained through clinical testing or other means to be de-identified and linked to health records for sale or transfer to a third party for research and development purposes (eg, novel drug targets, patient and provider tools for managing health care). Unlike many other elements within the de-identified data set, however, the patient's genetic information in various forms (eg, DNA sequence, RNA sequence, aggregated variant data, optical mapping) may be sufficiently information-rich to permit reidentification of the patient using informatics tools in some cases and is considered by some to be inherently identifiable. Several commercial entities have developed business models in which the pecuniary value of patient samples and data collected in the delivery of care are incorporated into the transaction so that samples and data may be used directly by the company in the discovery process, sold for reasons of profit, or to offset testing costs to patients.

The Board of Directors of the American College of Medical Genetics and Genomics approved this statement on October 19, 2021.
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As a leader in advancing genomic medicine, the American College of Medical Genetics and Genomics (ACMG) is committed to its mission to educate the public on the benefits and risks of genomic testing as well as to protect and preserve patients' welfare, autonomy, and privacy. ACMG affirms the need for genomic research and discovery for the improvement of health outcomes and patient experience. The exchange of de-identified genomic data, however, represents a potential avenue for breach of privacy that is inadequately protected by prevailing regulations and norms of practice. Consequently, there is a compelling need to establish standards of practice around the sale, transfer, or exchange of human clinical and genetic information to ensure the welfare of patients and community members in terms of privacy, consent, and the public's trust within the health care enterprise and the delivery of care. In developing a policy to address these practices involving use of genetic information, the term *stewardship* best captures this goal. Stewardship is a concept within the domain of professionalism that conveys a responsibility to deal faithfully with something that is entrusted to one's care or oversight.

Therefore, ACMG has established this policy to define the following as key elements related to the use, transfer, and sale of de-identified patient or consumer data obtained through the process of screening or diagnostic testing.

- When ordering genetic tests, clinicians should alert patients of any laboratory policies noted in the laboratory's consent/requisition form about how patient results and data may be shared in de-identified form for research, giving patients the choice to participate or not. At a minimum, patients must have the opportunity to opt out.
- The testing laboratory is the primary steward over the protection of the patient's interests in controlling the privacy of genetic information.
- In their stewardship of such data, laboratories should consider whether particularly sensitive uses, such as use of data to study genetic contributions to psychosocial traits, merit additional protections.
- A Data Use Agreement should specify use and transfer limitations; these must be in place to safeguard consumers from being exploited.
- Resale or other secondary transfers of human genomic data should not be permitted without appropriate authorization and an agreement between the parties that the data will not be reidentified; these same obligations for patient protection would accrue to any entity that acquires a laboratory and its assets or who gains access to genomic records by other means.
- Reidentification of individuals apart from their express permission is not permissible without their informed consent or authorization.
- Genomic sequencing of an individual without their consent or knowledge constitutes an invasion of privacy and is inappropriate except where laws permit specific applications (eg, law enforcement).
- An individual's decision whether or not to permit their data to be shared for research or discovery should not impede their access to routine clinical testing in the health care setting. For specific tests that are excluded from a patient's insurance coverage and for which a sponsor may offer no-cost investigational testing, opt-in strategies with informed consent should be used.

While this policy is primarily directed toward the sharing of genetic data and samples received by a clinical laboratory with outside entities, the College encourages full transparency with patients and consumers on the data obtained from samples (eg, genomic sequencing) and the many ways that patient samples and data are integrated into laboratory processes, such as quality assurance, test development, and knowledge sharing through public genomic data repositories such as National Institutes of Health–sponsored ClinVar,[1] which does not require explicit patient consent.[2]

Genetic and genomic data are complex and tightly linked both to the identities of individuals and to their health and disease states. Researchers, clinical laboratories, businesses, and other institutions have an ethical obligation to recognize the potential for breaches of privacy in their quest for medical advances tied to genomics-informed discovery. As technology, medicine, law, and society continue toward inclusion of genomics into health care and pursuit of the public good, it is incumbent upon all who are engaged in care and discovery to maintain a respect for the persons from whom these data are received. We must eschew personal and institutional conflicts of interest as business models evolve in order to preserve and advance trust and confidence in our professional communities as we serve the common good. These principles should be applied and reexamined frequently, even as the field and landscape of genomic medicine are changing.

Conflict of Interest

All authors declare no conflicts of interest.

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